ABSTRACT

A method for the diagnosis of SMEI in a patient comprising:

(1) detecting an alteration in the SCN1A gene, including in a regulatory region of the gene, in a patient sample;

(2) ascertaining whether the alteration is known to be SMEI associated or non-SMEI associated; and

(3) (a) establishing a diagnosis of a high probability of SMEI where the alteration is known to be SMEI associated; or

(b) establishing a diagnosis of a low probability of SMEI where the alteration is non-SMEI associated; or

(e) or, if not known to be either,

(i) considering genetic data for parents and/or relatives;

(ii) establishing whether the alteration has arisen *de novo* or is inherited; and

(iii) establishing a diagnosis of a low probability of SMEI where the alteration is inherited but a diagnosis of a high probability of SMEI if the alteration is *de novo*.